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REPROMEDA BIOLOGY PARK

Detecting Chromosomal Anomalies in Early Embryos through Preimplantation Genetic Screening (PGS)

By Kateřina Veselá

Reproduction genetics is continuously becoming one of the most important focus areas of assisted reproduction centres. Although it may not seem obvious, infertility, particularly infertility caused by advanced maternal age, is very closely linked to genetic factors. Numerical chromosomal anomalies, so called aneuploidies, arising in female oocytes, are one of the most serious causes of infertility. Many studies have confirmed that the number of aneuploidies occurring in oocytes increases significantly with advancing female age. These aneuploidies can be detected in early embryos through preimplantation genetic screening (PGS). In addition to this, PGS methods are currently not limited only to aneuploidy screening. Assisted reproduction centres are now able to provide help for clients not only with fertility problems, but also for couples with chromosomal translocations (often resulting in miscarriage or high-risk pregnancies), hereditary diseases or predisposition to cancer.

PGS, which is carried out in the course of the IVF treatment, allows us to transfer perspective embryos with a normal chromosomal set, i.e. euploid embryos, into the uterus. These embryos have a higher implantation potential, a lower abortion rate, and hopefully an overall higher chance of leading to the birth of a healthy offspring.

Incorrect chromosomal numbers in embryos are one of the most significant causes of embryo implantation failures and of unsuccessful IVF cycles in general. Most types of aneuploidies are not compatible with life, although in specific cases, chromosomal abnormalities, may lead to live births of affected foetus. The most common and well known conditions caused by chromosome aneuploidies include Down syndrome (extra chromosome 21), Patau syndrome (extra chromosome 13), or Edward syndrome (extra chromosome 18). However, the majority of these pregnancies end with foetal abortions (either spontaneous or induced).

PGS aims at improving pregnancy and live birth rates by embryo screening for chromosomal abnormalities. Screening is conducted in the shortest time and by the safest therapeutic means possible. Other advantages of PGS include:

- Reduction rate in miscarriages
- Decrease in the rate of pregnancy complications
- Treatment time and cost reduction, via decreasing the number of cycles necessary

- Multiple pregnancies reduction, due to one-embryo-only transfer policies
- Providing crucial diagnostic information, allowing the patient to decide whether to undergo another IVF cycle or consider gamete donation.

Without PGS, embryos for uterus transfer are selected solely on their visual quality, morphology, and growth dynamics. Regrettably, these criteria are not sufficient to exclude embryos with genetic abnormalities.

Due to preimplantation screening, we can inspect genetic qualities of selected embryos and consequently choose an embryo suitable for embryo transfer. This morphological and genetic selection ensures an increased embryo implantation potential and a decreased abortion rate. Therefore, couples with a poor chance of IVF success, can significantly improve their odds and their embryo implantation potential by genetic screening. These couples then have the same chance of embryo implantation as other couples, which is more than 40% for a single embryo transfer.